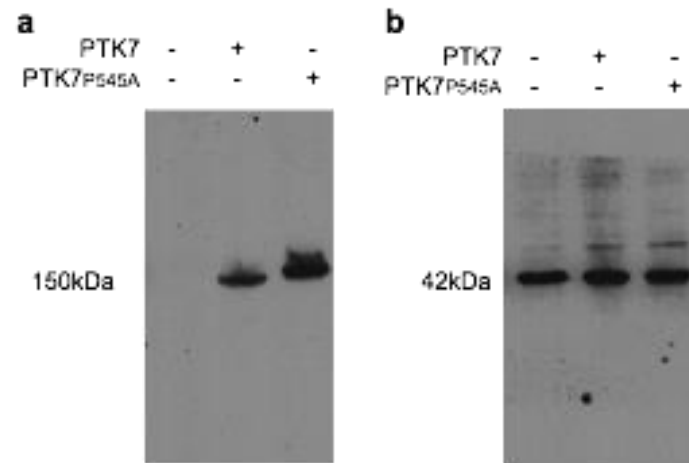


Supplementary Figure 1



Full western blots for images depicted in Figure 4. Full blots for (a) PTK7 and (b) actin using total cell lysates made from wild-type (WT) shield stage embryos that were injected at the one cell stage with 250 pg of either wild-type *PTK7* or *PTK7*^{P545A} mRNA. Both blots were made following transfer from the same protein gel.

Supplementary Table 1.

EXON	Chr 6 BP Position	SNP	Major allele/ Minor allele	Coding Type	AA Change	AA Position	cDNA Position	GERP	IS allele frequency	NHLBI-ESP EA allele frequency	NHLBI-ESP AA allele frequency
exon5	43098298	rs140093109	G/A	synonymous	NO	237	711		1/192	1/8600	2/4406
exon7	43100425	rs34021075	A/T	missense-near-splice	T->S	410	1228	5.01	4/192	174/8570	13/4384
exon11	43109421	N/A	C/G	missense	P->A	545	1633	5.46	1/192	N/A	N/A
exon12	43109751	rs6905948	G/A	synonymous	NO	617	1851		64/192	3262/8600	1975/4406
exon13	43110003	rs45453593	C/T	synonymous	NO	671	2013		2/192	69/8600	4/4406
exon15	43112267	rs34764696	C/T	missense	A->V	777	2330	3.67	10/192	604/8600	191/4406
exon16	43113047	rs202024669	G/A	synonymous	NO	839	2517		1/192	0/8600	2/4406
exon19	43127577	rs55921533	C/G	synonymous	NO	975	2925		1/192	73/8600	6/4406
exon20	43128519	rs34865794	G/A	missense	R->Q	1038	3113	4.26	6/192	89/8600	310/4406

PTK7 variants in IS samples. PTK7 coding exons were re-sequenced in 96 IS case samples. Numbering is in reference to transcript ID NM_002821.4/ENST00000230419. Results are given as allele frequencies alongside public results. NHLBI-ESP = National Heart Lung Blood Institute Exome Sequencing Project; EA = European American; AA = African American

Supplementary Table 2.

PTK7 Variant	Major allele	Minor allele	Affection status	Total genotyped	After QC	Allele counts		Individual P value *	Collapsed P value*
						Major allele	Minor allele		
rs34021075	A	T	control	755	753	1483	23	0.6195	0.3720
			case	1392	1378	2707	49		
rs34865794	G	A	control	755	749	1486	12	0.5102	
			case	1392	1350	2672	28		

Summary of genotyping PTK7 variants rs34021075 and rs34865794 in a follow-up cohort of non-Hispanic white (NHW) cases and NHW controls. Data shows the association of these two variants with IS is not statistically significant.

* P values were calculated using Fisher's exact test.